*Eco*RI restriction map (350 kb)

cen ↑   20 kb   ↑ tel

D14S990   Selected cDNA clones: D14S1457

GEHM-derived[3]

50 kb   candidate interval

217 kb

FIG. 1A   8kb *Hind* III subclone

Human *PAB II* genomic structure (6002 bp)

1   2   3   4   5   6   7

5'UTR   3'UTR

↑ intron 1

OPMD   intron 6

$(GCG)_n$ expansions   1 kb

▨ : ORF

FIG. 1B

FIG. 2A FIG. 2B FIG. 2C FIG. 2D FIG. 2E

FIG. 2F

(GCG)$_{6+n}$
expansions
N 2 3 4 5 6 7

I   V VIII   IX

GATC   FIG. 2G

$(GCG)_o$

←ATG

GATC GATC GATC

N   ht.   hm.
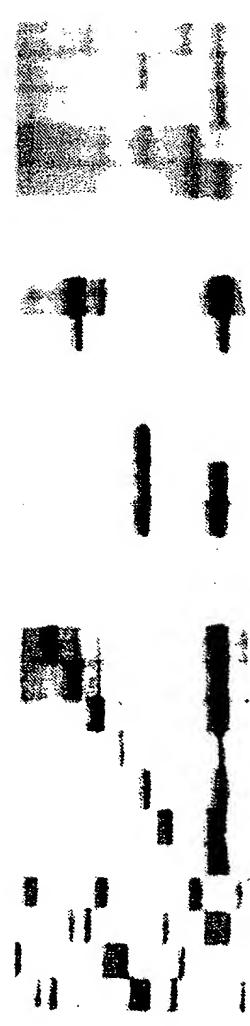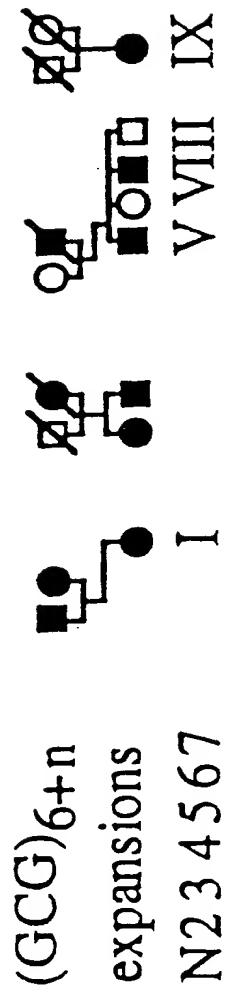
OPMD dominant mutations:

N: ATGGGCGGCGGCGGCGGCGGCGGCGGCAGCAGCA

ATGGGCGGCGGCGGCGGCGGCG(GCG)$_{2-7}$GCA

Polyalanine insertions:

N: MAAAAAAAGAAGAGGRGS

MAAAAAAA(A)$_{2-7}$AAAAGAAG

FIG. 3
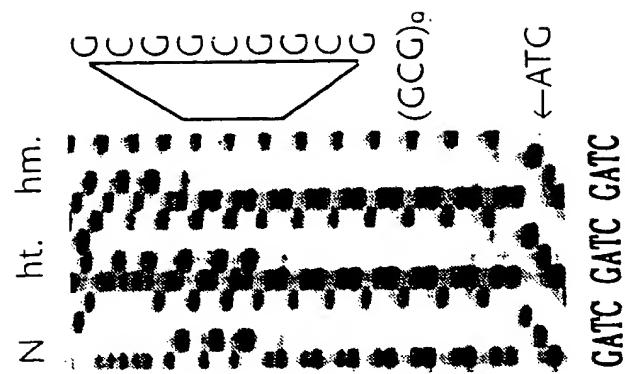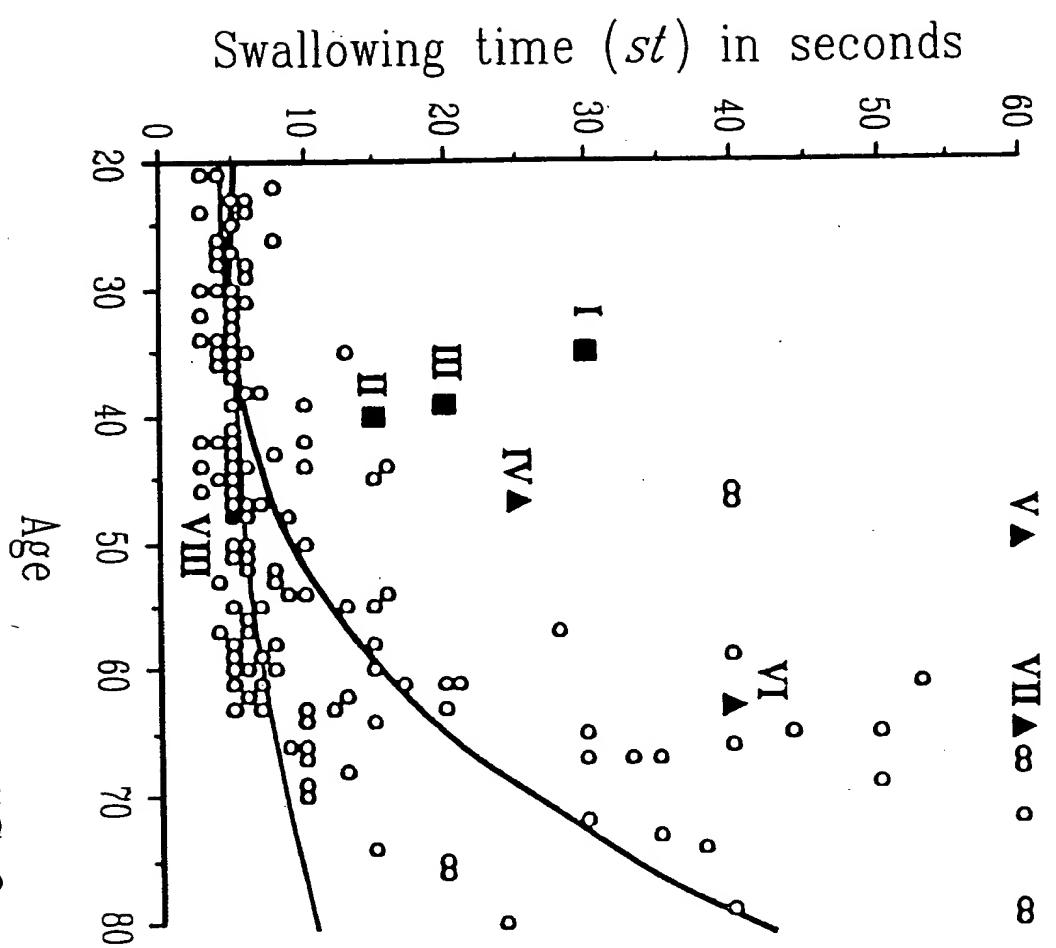
```
   1 aatgaaggtg gacacccaaa tagccccaat acaaatgcct gttcaatcaa ccaaacatct
  61 aagcagcaca tctatgtggt agcatattgc caggccgtga gactgcgaat ataaatagga
 121 accgcccctc atctgcaggc gctcacaacc tagttagcaa acagtaaaac aattaagcgc
 181 gccgtggaca taggcccact tgtcctggga aatgagggga agctggggtt tgcagtggtt
 241 tgattgaagg gggactacat gttagaggca cagactgggt gcaggtacac ccaaaggaac
 301 gagaagagtg gaaggaaaca acatccacaa agtaaccaca tgctggcgta tcgaaggccg
 361 tgatttacgg ttttgagact ttacctcgcc agcaaagggg ggccagtctg ttagcggtgc
 421 agattggagg ggtgacattg gaagctgtcc aggaaaaaga aaatggaact gactgaggaa
 481 aggcctacgc aagaggggcg gacagacagg acttgtgact agtagctctg gactgaggaa
 541 tcctccctgc tttctggtgc aatgcttcat tcacaacctc cattttcagc aacatcccat
 601 gaaagtaagc tccctcctgg aatgcttcat tcacaacctc cattttcagc aacatcccat
 601 gaaagtaagc tccctcctgg agatacaaag tgctgctctg tttggggcct
 661 ctactggtgc agatacaaag cagtagtatc aagatggcgg cccctagga
 721 caccggcca acagctcact agctggcaag cagtagtatc aagatggcgg cccctagga
 781 ctggctagtc atgtgacctc gggttttcca agtttgaagc ccggcagtc tttcgggggc
 841 aaggttcacc tgtcacgaaa cgagtgtcac cccttcgact ctcgcaagcc aatcggcatc
 901 tgagactggg ccactgcggt gaggcgatcg gaagattggt cctttccagt cgcctagcta
 961 gggccaatca cggagacgtc catacttcgc gggccgccc gtaggccggg gagaagcagg
1021 aatatcgtca cagcgtggcg gtattattac ctaaggactc gataggaggt gggacgcgtg
1081 ttgattgaca ggcagatttc cctaccggga tttgagaatt tggcgcagtg ccgccttag
1141 aggtgcgctt atttgattgc caagtaatat tcccaaatgg agtactagct catggtgacg
```

FIG. 4A

```
1201  ggcaagcagc ttgagctaat gagtcctccg tggccggcgc agctctccac atgccgggcg
1261  gcgggcccca gtctgagcgg cgatggcggc ggcggcagca ggcccagcag cagcgggggc
1321  tgcgggcggt cgggggctccg ggccggggcg gcggcgccat cttgtgcccg gggccggtgg
1381  ggaggccggg gaggggggcgc cggggggcgc agggaacggcc tggagtctga
1441  ggaactggag cctgaggagc tgctgctgga gcccgagccg gagcccgagc ccgaagagga
1501  gccgcccccgg cccccgggagc tccgggcccct gggcctggtt cgggagcccc
1561  cggcagccaa gaggaggagg actggtcgag ggtgaccccg gggacgcggc
1621  cattgaggac ccggtgagga gcgagcaggc cgcggcctgg cgcgtcactg
1681  gaggcccaga gctcggcgca gcggtggcag gcgggggtg gggttgggcg gggaataacg
1741  tggctgggc gggatcgggc cagcgatcac tacaaggggc ccgactggct
1801  tgattcgggc gtcacgggtg cctagtgttg ttctagagag ggtagctttt cttttatcac
1861  gacctcgca tgggcgagg gaaatggccg agcatggctg aggcgcgctc tggccgagag
1921  cagggcacag cccctgcgtt ggttcctctt aagctgtcct ccatacctc cccacttata
1981  ttaggagctg gaagctcgagt caggggagatg gaggaagaag ctgagaagct
2041  aaagagcta cagaacgagg tagagaagca gatgaatatg agtccacctc caggcaatgc
2101  tgagtaactg gcggttgcac gcggagcccg ggttctcggg ttgaagggt tgtgggggag
2161  atggggaatg tgggtttaga tactcggcac cctggagctg cttgtctgag ctattatgac
2221  tgtgccggcg tcatagtccg ctctgacctt tgtgaggcag aactgatatt
2281  ttggtggtgg tagcctttgtg cctcccttg ctctgttta attgtgttgc tctttattct
2341  tagtctacgt ctatctttct ttgcgtgctc gcatttgacc ttcaaatcta
```

FIG. 4B

```
2401 atagtttttc ctccaattgg agacgcttta ggattctaag agaaagcaag ctggaagggg
2461 tttcccctt aaattctaga aatgtggagt ctcagcccac ttaatttgc tcactcttaa
2521 aagcatttca accaaagcca ttcattaggg atttgatttg gaggcagga gggattccta
2581 tactgtttta agtgtgtatt aattctttca atttatcgaa ttattagtg agtaacctgc
2641 tatgcactag gcactattct cggcttgtgg gtacagcagg gaacagcaca gaccaaaatc
2701 tttgccttca ctgagcttat gggatagtgc tggtggtgga agtgcaacat attggtcaag
2761 tagaaaacaa gtgtgtggtt tttgtaaaaa attatttttt cctgatagct ggccggtga
2821 tcatgtccat tgaggagaag atggaggctg aagttcttct catctatgtt ggcaatgtga
2881 cgtactgggg ctctgactgg ggttgggggc aagtttctct tttggggaat tatttaatag
2941 tcctgaaaga acatctccgg gatagatgtg gttttgggtg tggagggagt gtgggaagga
3001 ggttaaaggt aatggaatga tcagtaatca gcaaaggctc tgggtttgga aggaaaagag
3061 attaattcct caaattacca gatttcatgt gctttggtgt atgatggccc agaccaaagg
3121 ctcgggaggg ttctttgag acaggaattt gcctggtgcc tgtgaaattt ttctccctc
3181 atcaggtgga ctatggtgca acagcagaag agctggaagc tcactttcat ggctgtggtt
3241 cagtcaaccg tgttaccata ctgtgtgaca aatttagtgg ccatcccaaa ggtaaagtaa
3301 aggggagtaa gttgagataa tttaaattac agtgtacaaa tagataaatt atgtttata
3361 ttgagcagta agttattgg tgttgatctgt gtcacctaa atgtcttcag aggccagata
3421 taatgttgat atatcaggag ttgcacctaa atgtcttcag aggccagata acaaaaatga
3481 aggctagatg tgggtgggat tacgaactag aaggggaggg gcagcttcta cttggcctat
3541 tatggcatat ggaaattcag gccctgtgtg tcttattttt acaaatttca aagagtagct
```

FIG. 4C

```
3601 ggaaattta aaatttaaat gattcgaat ttccatttag aagaattttg
3661 acaaataaaa aatataactg cattgtagcc caaaacgaag ggttgaatt
3721 gacctgtgag gtatttgtaa cctcagagag aattcttttc aggtttgcgt
3781 atatagagtt ctcagacaaa gagtcagtga ggccttagat gagtccctat
3841 ttagaggaag gcaaatcaag gtaagcctat gttccattgct tgtctagttg tgtataaact
3901 ctccaggttg cctttaaggc tatcatttgt ctcaggtgat cccaaaacga
3961 accaacagac caggcatcag cacaaacagac cgggggtttc cacgagcccg ctaccgcgcc
4021 cggaccacca actacaacag ctcccgctct cgattctaca gtggtttaa cagcaggccc
4081 cgggggtcgcg tctacaggtc aggatagatg ggctgctcct ctttcccccg cctcccgtga
4141 gccccgtatg cttcctcctc tctgcagt ggaacctccc tccccacc cctcccgtg
4201 gtcttcagga actttgtctc ctgcctgtgc aggtagttgc aggccaggcc
4261 agaaggcagc ctcatcatct tttctgcagt agaaattggt gataaggct gcatcctcc
4321 cttggttcaa agaggcttcc accccagcc ttttttct tgggagttgg tggcattga
4381 aggtgtttgc ggaggaaca gggaggcccag gaagttgaaa gcactgcttg
4441 gacatttgtt acttttttcg gagttaggga actgaactc cccttggaaga
4501 ataccagagg ctagctagtt gatccttcca acagccttgt gggaggatt tgagatactt
4561 attctttatt tgacccagtc ttgcaaggtt aacttctcac tgggcctagt gtggtnccca
4621 ggttttttgcc ttgcttcact tctgtctcta catttaaata gacgggttag gcatataaac
4681 cttggctttt cataagctct acctgcctat cccaggagt taggaggat ctattgtga
4741 aggccctagg gtttaaaaac tgtggaggac tgaaaaactg gataaaaagg gggtcctttt
```

FIG. 4D

```
4801 ccttgcccct gtctctcact cagatgcgct tctttttcgc cactgtttgg caaagttttc
4861 tgttaagccc ccctcccct gcccagttc tccaggtgc gttactatt ctgggatcat
4921 gggtcggtt ttaggacact tgaacacttc ttcccttcac agtaactggg
4981 gcagggggcct acgggggagg gcttgtactg tgatcacgtt aacacctaac
5041 tctccttctt tcttccaggg gccgggctag agcgacatca tggtattccc cttactaaaa
5101 aaagtgtgta ttaggaggag agagaggaaa aaaagagggaa agaaaaagaat
5161 taaaaaaaaa aaaaagaaaa acagaagatg accttgatgg aaaaaaaata tttttaaaa
5221 aaaagatata ctgtggaagg gggagaatc ccataactaa ctgctgagga gggacctgct
5281 ttgggagta gggggaaggcc caggggtgg tgcttgccca tgctttccctg tttttccccag
5341 tcgccatgga cacgtctcaa ctgcgcaagc tgcttgccca gtttccctg cccccttcac
5401 cccctgggc ctgctcaagg gtagtgggc gtggtggta ggaggtttt ttttacccag
5461 ggctctggaa ggacaccaaa ctgtctct tgttaccttc cccacccctc tctcctcgcc
5521 tttcacagtc ccctcctgcc tccagattgc cagccaagtc taccaccac ccccctcgcc
5581 tttctccggc tccctgcccc ctggtgatct agccggaaga tctttgttcc
5641 tcttttctg tttgagtgt aggtttctgt atttttgttc ctttgttgtt
5701 gctccagcg aaattcccct tccccctggg gaaatgcact accttgtttt
5761 ggggggttta gggggtgtttt ttgttttttcag ttgttttgtt ttttttttcc
5821 tttgccttt ttccctttta tttggaggga atgggaacag ggaggtggga
5881 ggtggattt gtttattttt ttagctcatt tccaggggtg tttaatatgt
5941 gtcatgaata aagttgtttt tgaaaataaa ggaattttt tttaatatgt
6001 aa
```

FIG. 4E